MATR3 gene

matrin 3

Normal Function

The *MATR3* gene provides instructions for making a protein called matrin 3, which is found in the nucleus of the cell as part of the nuclear matrix. The nuclear matrix is a network of proteins that provides structural support for the nucleus and aids in several important nuclear functions.

The function of the matrin 3 protein is unknown. This protein can attach to (bind) RNA, which is a chemical cousin of DNA. Some studies indicate that matrin 3 binds and stabilizes a type of RNA called messenger RNA (mRNA), which provides the genetic blueprint for proteins. Matrin 3 may also bind certain abnormal RNAs that could lead to nonfunctional or harmful proteins, thereby blocking the formation of such proteins. Other studies suggest that the matrin 3 protein may be involved in cell survival.

Health Conditions Related to Genetic Changes

amyotrophic lateral sclerosis

distal myopathy 2

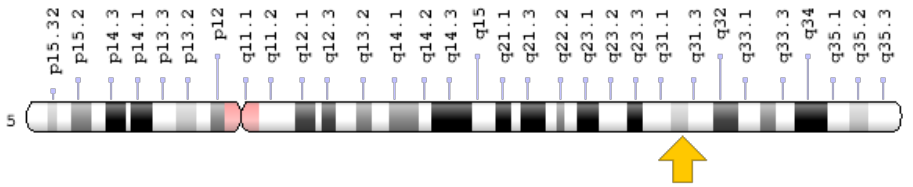
At least one mutation in the *MATR3* gene has been identified in people with distal myopathy 2, a condition characterized by muscle and vocal cord weakness. The *MATR3* gene mutation associated with distal myopathy 2 changes a single protein building block (amino acid) in the matrin 3 protein. This mutation, known as Ser85Cys (or S85C), replaces the amino acid serine with the amino acid cysteine at position 85 of the protein.

The effect of the S85C mutation on the function of the matrin 3 protein is unknown, although one study indicates that the mutation may change the location of the protein in the nucleus. Researchers are working to determine how this gene mutation leads to the signs and symptoms of distal myopathy 2.

Chromosomal Location

Cytogenetic Location: 5q31.2, which is the long (q) arm of chromosome 5 at position 31.2

Molecular Location: base pairs 139,273,752 to 139,331,677 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MATR3_HUMAN
- matrin-3
- matrin-3 isoform a
- matrin-3 isoform b
- MGC9105
- MPD2

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (2nd Edition, 2000): Functional Domains Within the Nucleus
  https://www.ncbi.nlm.nih.gov/books/NBK9915/#A1354

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MATR3%5BTIAB%5D%29+OR+%28matrin+3%5BTIAB%5D%29%29+OR+%28%28matrin-3%5BTIAB%5D%29+OR+%28MPD2%5BTIAB%5D%29+OR+%28VCPDM%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- MATRIN 3
  http://omim.org/entry/164015

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MATR3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MATR3%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6912
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9782
- UniProt
  http://www.uniprot.org/uniprot/P43243

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